Amendments to the Claims

Please cancel Claims 3-8, 12, 15, 19-30, 31 and 33-36 without prejudice. Applicants reserve the right to prosecute such claims or the subject matter thereof in further applications. Please amend claims 1 and 32. Please add new claims 40-52. The Claim Listing below will replace all prior versions of the claims in the application:

Claim Listing

1. (Currently amended) An isolated human PAB II gene comprising a polymorphic GCG repeat in exon I thereof, wherein said polymorphic GCG repeat has the sequence

ATG $(GCG)_{6+n}$ GCA,

~~with n being selected from 1 to 7 and wherein said polymorphic repeat of said GCG repeat is indicative of a disease in a human patient~~,wherein n is selected from the group consisting of:

a) n=0, wherein said gene is associated with a non-disease phenotype; and

b) n is selected from 1 to 7, wherein said gene is associated with a phenotype of oculopharyngeal muscular dystrophy, selected from at least one of protein accumulation in a cell nucleus, swallowing difficulty, and ptosis, and

wherein said human PAB II gene comprises the sequence as set forth in SEQ ID NO: 18, excluding nucleotide positions 1283 to 1306 thereof, said nucleotide positions 1283 to 1306 being replaced by said ATG $(GCG)_{6+n}$ GCA sequence.

2-8. (Canceled)

9. (Previously presented) An isolated nucleic acid sequence comprising a polymorphic GCG repeat of exon I of a human PAB II gene, wherein said polymorphic GCG repeat has the sequence

ATG $(GCG)_{6+n}$ GCA,

with n being selected from 1 to 7 and wherein said polymorphic repeat of said GCG repeat in a patient=s human PAB II gene is indicative of a disease in said human patient.

10. (Canceled)

11. (Previously presented) The nucleic acid sequence of claim 9, wherein n is selected from 2 to 7, and wherein said polymorphic repeat of said GCG repeat is associated with an increased severity of said disease.

12. (Canceled)

13. (Previously presented) A method for the diagnosis or prognosis of oculopharyngeal muscular

dystrophy (OPMD), a disease associated with protein accumulation in a cell nucleus, and/or swallowing difficulty and/or ptosis in a human patient, which comprises:

a) obtaining a nucleic acid sample of said patient; and

b) determining allelic variants of a GCG repeat in exon I of the PAB II gene, said GCG repeat having the sequence

$$ATG\ (GCG)_{6+n}\ GCA,$$

wherein n is selected from 0 to 7, and
whereby at least one of the two alleles of said GCG repeat having an n equal to 1 to 7, is indicative of OPMD.

14. (Original) The method of claim 13, wherein n is selected from 2 to 7, and wherein said allelic variant is associated with an increased severity of said disease.

15. (Canceled)

16. (Previously presented) The method of claim 13, wherein said first allele of said GCG repeat has an n which is equal to 1.

17. (Previously presented) The method of claim 16, wherein said second allele of said GCG repeat has an n selected from 2 to 7, and wherein said first allele is a modulator of the severity of the phenotype associated with said second allele.

18-31. (Canceled)

32. (Currently amended) The human PAB II gene of claim ~~31~~, wherein n=0, and wherein the sequence of said gene is as set forth in SEQ ID NO:18.

33-36. (Canceled)

37. (Previously presented) An isolated PAB II nucleic acid sequence comprising a polymorphic GCG repeat having the sequence

$$ATG\ (GCG)_{6+n}\ GCA,$$

wherein n is selected from the group consisting of:

a) n=0, wherein said nucleic acid sequence is associated with a non-disease phenotype; and

b) n is selected from 1 to 7, wherein said nucleic acid sequence is associated with a phenotype of oculopharyngeal muscular dystrophy, selected from at least one of protein accumulation in a cell nucleus, swallowing difficulty, and ptosis.

38. (Previously presented) The isolated nucleic acid sequence of claim 37, wherein n=0, and

wherein said sequence comprises the sequence as set forth in SEQ ID NO:18.

39. (Previously presented) The isolated nucleic acid sequence of claim 37, wherein n=0, and wherein said sequence comprises the sequence as set forth in SEQ ID NO:1.

40. (New) The isolated nucleic acid sequence of claim 37, wherein n=0, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:2.

41. (New) The isolated nucleic acid sequence of claim 37, wherein n=1, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:3.

42. (New) The isolated nucleic acid sequence of claim 37, wherein n=2, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:4.

43. (New) The isolated nucleic acid sequence of claim 37, wherein n=3, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:5.

44. (New) The isolated nucleic acid sequence of claim 37, wherein n=4, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:6.

45. (New) The isolated nucleic acid sequence of claim 37, wherein n=5, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:7.

46. (New) The isolated nucleic acid sequence of claim 37, wherein n=6, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:8.

47. (New) The isolated nucleic acid sequence of claim 37, wherein n=7, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:9.

48. (New) A method for the diagnosis or prognosis of oculopharyngeal muscular dystrophy (OPMD), a disease associated with protein accumulation in a cell nucleus, and/or swallowing difficulty and/or ptosis in a human patient, which comprises:

a) determining allelic variants of a GCG repeat in exon I of the PAB II gene from a sample of a patient, said GCG repeat having the sequence

$$ATG\ (GCG)_{6+n}\ GCA,$$

wherein n is selected from 0 to 7, and
whereby at least one of the two alleles of said GCG repeat having an n equal to 1 to 7, and is indicative of OPMD.

49. (New) The method of claim 48, wherein n=0, and wherein said GCG repeat has the sequence set forth in SEQ ID NO:2.

50. (New) The method of claim 48, wherein said first allele of said GCG repeat has an n which is equal to 1, and has the sequence set forth in SEQ ID NO:3.

51. (New) The method of claim 48, wherein n is selected from 2 to 7, wherein said allelic variant is associated with an increased severity of said disease, and wherein said GCG repeat has the sequence selected from the group consisting of:

a) SEQ ID NO:4, when n=2;

b) SEQ ID NO:5, when n=3;

c) SEQ ID NO:6, when n=4;

d) SEQ ID NO:7, when n=5;

e) SEQ ID NO:8, when n=6; and

f) SEQ ID NO:9, when n=7.

52. (New) The method of claim 50, wherein said second allele of said GCG repeat has an n selected from 2 to 7, said first allele is a modulator of the severity of the phenotype associated with said second allele, and wherein said GCG repeat of said second allele has the sequence selected from the group consisting of:

a) SEQ ID NO:4, when n=2;

b) SEQ ID NO:5, when n=3;

c) SEQ ID NO:6, when n=4;

d) SEQ ID NO:7, when n=5;

e) SEQ ID NO:8, when n=6; and

f) SEQ ID NO:9, when n=7.